

## Bart's Syndrome with Unilateral Choanal Atresia

Fairy Susan Varghese<sup>1</sup>, Sunil K Agarwalla<sup>2</sup>, Geetanjali Sethy<sup>3</sup>, Pranab K Panigrahi<sup>4</sup>

### ABSTRACT

**Introduction:** Bart's syndrome is clinically described as the association of congenital localized absence of skin (CLAS), epidermolysis bullosa (EB) and nail abnormalities particularly dystrophy. CLAS is now regarded as a manifestation of EB. This familial autosomal dominant disorder was first described in 1966 by Bart. We report a neonatal case of Bart's syndrome in view of the exceedingly rare nature of the entity with an atypical finding of unilateral choanal atresia.

**Case Report:** A male newborn baby was brought with absence of skin and extensive peeling on the dorso lateral aspects of left leg and plantar surface of left foot. A naso gastric tube could not be passed from the left nostril to the pharynx suggesting unilateral choanal atresia. On day-3 of life dystrophic nail changes appeared and ultimately the child died due to sepsis.

**Conclusion:** In newborns with congenital absence of the skin in the presence of bullae formation, Bart's syndrome should be considered and septicaemia should be anticipated as a complication.

**Key Words:** Bart, choanal atresia, Congenital localized absence of skin (CLAS), epidermolysis bullosa (EB), septicaemia

### INTRODUCTION

The approach to a child with vesiculobullous disease is a complicated and a challenging task given the vast differential diagnosis and can be a cause of great concern to the parents and the physician alike. Not only can the appearance be upsetting and the blisters tender, but also the development of a number of childhood vesiculobullous disorders may be the harbinger of a life of disability and discomfort and may be potentially fatal.<sup>1</sup>

Bullae may arise as a manifestation of common conditions that are not generally regarded as blistering disorders such as acute eczema, cellulitis and insect bite where the intensity of the inflammatory process is sufficient to cause the epidermis to separate from the dermis.

In 1966 and 1971, Bart and co-workers<sup>2,3</sup> described a large kinship in which 26 members were found to have at least 2 of the following traits (a) localized absence of skin (b) blistering of the skin with/without mucous membrane involvement resembling epidermolysis bullosa and (c) congenital absence and/or dystrophy of the nails.

Aplasia cutis restricted to the lower extremities was noted in the newborn period. Recurrent blistering and at times shedding of the nails began in infancy or childhood. Both the aplasia cutis and the blistering lesions healed without scarring. Moreover, in the kinship reported by Bart, the pattern

of inheritance was clearly autosomal dominant with variable penetrance.

Recent studies have further classified Bart's syndrome as a variant of epidermolysis bullosa. Here we report a case of Bart's syndrome with a rare association of unilateral choanal atresia who succumbed on day 15 of life.

### CASE REPORT

A male newborn, born by normal vaginal hospital delivery, the 3<sup>rd</sup> order child of a non consanguineous marriage revealed absence of skin and extensive peeling on the dorsolateral aspects of left leg (Figure 1) and plantar surface of left foot (Figure 2).

There were multiple blisters on the dorsum of trunk and buttocks (Figure 3); no lesions in the oral cavity and nails. He was the 3<sup>rd</sup> order child of a non consanguineous marriage. The mother gave a history of 2 post natal deaths on days 5, 7 with similar skin lesions and home delivery in both cases. She had improper antenatal care in all 3 deliveries with no antenatal scans done.

A naso gastric tube could not be passed from the left nostril to the pharynx suggesting unilateral choanal atresia. The birth weight, length and head circumference were 2.5kg, 49cm, 34cm respectively and were appropriate for age.

Complete blood count revealed leukocytosis and gram staining of blister showed gram positive cocci. Appearance of dystrophic and hypoplastic nail changes occurred on the third day (Figure 4). From day-5 of hospitalisation a gradual increase in levels of blood urea nitrogen (BUN) and creatinine were detected. Echocardiography, cranial and abdominal ultrasounds were normal. A skin biopsy could not be done due to parents' refusal. The patient was managed conservatively with sterile skin care, total parenteral nutrition with umbilical catheter, and antibiotics but on day-15 the child succumbed due to suspected septicaemia and acute renal failure.

### DISCUSSION

The first case report of Bart's syndrome originated in the year 1966 in a family with manifestations of extensive blistering of the mucous membrane and skin, nail dystrophy and congenital

<sup>1,4</sup>Junior Resident, <sup>2,3</sup>Associate Professor, Department of Pediatrics, MKCG Medical College, Brahmapur, Odisha-760004, India.

**Corresponding author:** Dr. Fairy Susan Varghese, Junior resident-3, Room No.113, PG Ladies Hostel 2, MKCG Medical College, Brahmapur, Odisha-760004, India.

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**Figure-1:** Absence of skin with extensive peeling on the dorso-lateral aspects of left leg, foot



**Figure-3:** Blisters on the dorsum of trunk, buttocks



**Figure-2:** Plantar aspect of left foot



**Figure-4:** Nail dystrophy on Day-3 of life

localised absence of skin (CLAS) on the lower leg. The exact cause of aplasia cutis congenital (ACC) in association with EB is not known. The assumption is that in utero blistering and erosions may be due to foetal movements leading to mechanical trauma.

The most commonly involved parts in CLAS are the extremities particularly the limbs and less commonly the parietal and occipital regions of the scalp. However the symmetry of involvement, well defined borders and involvement of the soles and toe webs are pointers against the explanation that in utero physical trauma causes the entity.

Zelickson et al analyzed Bart's kindred and demonstrated poorly formed anchoring fibrils and cleavage below the lamina densa on ultrastructural analysis. Genetic linkage studies mapped the gene for the disease in this family to chromosome 3p at or near the site of the gene encoding type VII collagen (COL7A1). Christiano et al performed mutation analysis in this family by DNA sequencing, which resulted in glycine to arginine substitution within the triple helical domain of type VII collagen in the affected individuals.

Epidermolysis bullosa (EB) can be of three subtypes based on the location of the bullae. They are as follows;

- EB simplex: In this type the bullae are located within the epidermis in the suprabasal area.
- Junctional EB: Here the bullae are sup epidermal located just above the basement membrane which has PAS staining properties.
- Dystrophic EB: In this variety the bullae are sup epidermal like the junctional type but are situated below the basement membrane<sup>4</sup>.

Although Bart's syndrome can be associated with any of the three subtypes of EB, it is most commonly associated with the dystrophic variety.

The cases originally described by Bart had also the dystrophic variety of EB as an association. A rare and severe variant is the Herlitzform variant of junctional EB. It is caused by mutations in the anchor protein laminin 513 and is usually lethal.

#### **Aplasia Cutis Congenita (ACC)**

In this entity there is true absence of skin in a localised or widespread pattern at birth. Scalp is the most common site of

involvement and other developmental anomalies may be associated. According to Frieden, the condition is classified into the following 9 categories.<sup>4</sup>

### Epidemiology

The incidence of ACC is around 3 cases per 10,000 newborns. There are no sexual or racial predilections. While most cases are sporadic, there have been familial cases reported in Frieden groups 1, 2,5,6,7 and 9. The clinical findings are present at birth. In some cases there may be a family history of a similar disorder. The discussion below follows Frieden's classification<sup>5</sup>.

**GROUP 1: Scalp Without Multiple Anomalies** - Scalp is the most common site of involvement. Nearly 86% of all solitary lesions occur on the vertex of the scalp near the parietal hair whorl. By definition, infants with Grp 1 ACC do not have multiple anomalies but patients with isolated anomalies are included in this group. Eggs are cleft palate, trachea-esophageal fistula, double cervix and uterus, congenital heart disease and omphalocele.

**GROUP 2: Scalp with limb anomalies (adams oliver syndrome)** - In this distinct subtype, distal limb reduction anomalies are found in association with solitary midline scalp defects. Inheritance is autosomal dominant.

**GROUP 3: Scalp with epidermal and sebaceous naevi**

**GROUP 4: Overlying embryologic malformations** - In various types of major malformations, there may be a congenital absence of skin overlying the deeper defect. Eggs include meningomyelocele, gastroschisis, porencephaly, etc

**GROUP 5: Associated with fetus papyraceus or placental infarct** - This is associated with the presence of a fetus papyraceus, which is found at delivery in the placenta and is caused by the death of a twin foetus during the second trimester.

**GROUP 6: Associated with epidermolysis bullosa (bart's syndrome)** - Here ACC usually occurs in the lower extremities with EB. Frieden proposed that the lesions develop on the lower limbs because of mechanical trauma in utero due to foetal kicking which leads to in utero blistering and subsequent erosions.

**GROUP 7: Extremities without EB**

**GROUP 8: Teratogens:** Few cases of ACC are linked to in utero infections with herpes simplex virus or varicella and to the medications valproate and methimazole given during pregnancy.

**GROUP 9: Associated with syndromes of malformation** - Examples is trisomy 13,4p syndrome, ectodermal dysplasia, focal dermal hypoplasia (Goltz syndrome).

### Pathophysiology and Histogenesis

The histology varies according to the depth of the aplasia and its duration. At birth, deeply ulcerated lesions may show complete absence of skin. After healing, the epidermis, if present, is thinned and flattened and there may be a proliferation of fibroblasts within the connective tissue stroma. Bullous ACC may show fibrovascular or oedematous stroma similar to the histopathological findings of encephaloceles and meningoceles, supporting the theory that ACC is a forme frusta of neural tube defects.<sup>6</sup>

Because ACC is a clinical finding that occurs in more than one disease state, it is very likely that there is more than one mechanism of pathogenesis. The disruption of skin development occurs in utero genetic factors are one mechanism by which the abnormality may be produced. Other proposed mechanisms include trauma, compromised cutaneous vasculature and teratogens.

### Differential Diagnosis

Diagnostic considerations include an encephalocele or a dermoid cyst. ACC must also be differentiated from the erosions of EB; this can be confusing since patients with EB can also have ACC. Other considerations include neonatal herpes, focal dermal hypoplasia and amniotic band syndrome.

### Therapy and Prognosis

Treatment is rarely necessary in more superficial lesions because the erosions and ulcerations almost always heal spontaneously with excellent prognosis. Occasionally patients require skin grafting.

Septicaemia, electrolyte imbalance, protein loss, and failure to thrive complicate the severe exudative skin lesions, often leading to death in these cases.

In newborns with congenital absence of the skin in the presence of bullae formation, Bart's syndrome should be considered, and the newborns should be examined in terms of developmental defects. Genetic counselling for affected families is extremely important for diagnosis of this rare familial disorder.

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